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NEWS 1		Web Page URLs for STN Seminar Schedule - N. America
NEWS 2		"Ask CAS" for self-help around the clock
NEWS 3	SEP 09	CA/CAplus records now contain indexing from 1907 to the present
NEWS 4	DEC 08	INPADOC: Legal Status data reloaded
NEWS 5	SEP 29	DISSABS now available on STN
NEWS 6	OCT 10	PCTFULL: Two new display fields added
NEWS 7	OCT 21	BIOSIS file reloaded and enhanced
NEWS 8	OCT 28	BIOSIS file segment of TOXCENTER reloaded and enhanced
NEWS 9	NOV 24	MSDS-CCOHS file reloaded
NEWS 10	DEC 08	CABA reloaded with left truncation
NEWS 11	DEC 08	IMS file names changed
NEWS 12	DEC 09	Experimental property data collected by CAS now available in REGISTRY
NEWS 13	DEC 09	STN Entry Date available for display in REGISTRY and CA/CAplus
NEWS 14	DEC 17	DGENE: Two new display fields added
NEWS 15	DEC 18	BIOTECHNO no longer updated
NEWS 16	DEC 19	CROPU no longer updated; subscriber discount no longer available
NEWS 17	DEC 22	Additional INPI reactions and pre-1907 documents added to CAS databases
NEWS 18	DEC 22	IFIPAT/IFIUDB/IFICDB reloaded with new data and search fields
NEWS 19	DEC 22	ABI-INFORM now available on STN
NEWS 20	JAN 27	Source of Registration (SR) information in REGISTRY updated and searchable
NEWS 21	JAN 27	A new search aid, the Company Name Thesaurus, available in CA/CAplus
NEWS 22	FEB 05	German (DE) application and patent publication number format changes
NEWS 23	MAR 03	MEDLINE and LMEDLINE reloaded
NEWS 24	MAR 03	MEDLINE file segment of TOXCENTER reloaded
NEWS 25	MAR 03	FRANCEPAT now available on STN

NEWS EXPRESS	MARCH 5 CURRENT WINDOWS VERSION IS V7.00A, CURRENT MACINTOSH VERSION IS V6.0b(ENG) AND V6.0Jb(JP), AND CURRENT DISCOVER FILE IS DATED 3 MARCH 2004
NEWS HOURS	STN Operating Hours Plus Help Desk Availability
NEWS INTER	General Internet Information
NEWS LOGIN	Welcome Banner and News Items
NEWS PHONE	Direct Dial and Telecommunication Network Access to STN
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FILE 'HOME' ENTERED AT 19:17:32 ON 10 MAR 2004

=> file biosis
COST IN U.S. DOLLARS

	SINCE FILE ENTRY	TOTAL SESSION
FULL ESTIMATED COST	0.21	0.21

FILE 'BIOSIS' ENTERED AT 19:17:55 ON 10 MAR 2004
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FILE COVERS 1969 TO DATE.
CAS REGISTRY NUMBERS AND CHEMICAL NAMES (CNs) PRESENT
FROM JANUARY 1969 TO DATE.

RECORDS LAST ADDED: 3 March 2004 (20040303/ED)

FILE RELOADED: 19 October 2003.

```
=> s pompe
           312 POMPE
           159 POMPES
L1         422 POMPE
                 (POMPE OR POMPES)

=> s l1 and (hexose (w) tetrasaccharide)
          7736 HEXOSE
          2001 HEXOSES
          9163 HEXOSE
                 (HEXOSE OR HEXOSES)
          1496 TETRASACCHARIDE
           396 TETRASACCHARIDES
          1773 TETRASACCHARIDE
                 (TETRASACCHARIDE OR TETRASACCHARIDES)
             0 HEXOSE (W) TETRASACCHARIDE
L2           0 L1 AND (HEXOSE (W) TETRASACCHARIDE)

=> s l1 and (tetrasaccharide)
          1496 TETRASACCHARIDE
           396 TETRASACCHARIDES
          1773 TETRASACCHARIDE
                 (TETRASACCHARIDE OR TETRASACCHARIDES)
L3           5 L1 AND (TETRASACCHARIDE)

=> d l3 kwic 1-5

L3   ANSWER 1 OF 5  BIOSIS  COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
TI   Analysis of a glucose tetrasaccharide elevated in Pompe
     disease by stable isotope dilution-electrospray ionization tandem mass
     spectrometry.
AB   Patients with glycogen storage disease type II (GSD II) typically excrete
     increased amounts of a glycogen-derived glucose tetrasaccharide,
     Glcalpha1-6Glcalpha1-4Glcalpha1-4Glc (Glc4), in the urine.  With the
     advent of a new enzyme replacement therapy for GSD II, there is a.  .  .
IT   .  .  .
        Metabolism; Methods and Techniques
IT   Parts, Structures, & Systems of Organisms
        plasma: blood and lymphatics; urine: excretory system
IT   Diseases
           Pompe disease: genetic disease, metabolic disease
        Glycogen Storage Disease Type II (MeSH)
IT   Diseases
```

glycogen storage disease type II: genetic disease, metabolic disease, therapy, GSD II
Glycogen Storage Disease Type II (MeSH)

IT Chemicals & Biochemicals
butyl 4-aminobenzoate derivatives; glucose; glucose **tetrasaccharide**: analysis; recombinant alpha-amylase

L3 ANSWER 2 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
TI Determination of oligosaccharides in **Pompe** disease by electrospray ionization tandem mass spectrometry.
AB. . . need for biochemical markers to monitor the efficacy of therapy and methods to quantify these markers in biologic samples. In **Pompe** disease, the concentration of a **tetrasaccharide**, consisting of four glucose residues, is reputedly increased in urine and plasma, but faster and more sensitive methods are required. . . spectrometry, of oligosaccharide concentrations in urine (n = 6), plasma (n = 11), and dried-blood spots (n = 17) from **Pompe**-affected individuals. Age-matched control samples of urine (n = 10), plasma (n = 28), and blood spots (n = 369) were also analyzed. Results: The mean **tetrasaccharide** concentration was increased in urine from infantile-onset (0.69-12 mmol/mol of creatinine) and adult-onset (0.22-3.0 mmol/mol of creatinine) **Pompe** individuals compared with age-matched controls. In plasma samples, an increased **tetrasaccharide** concentration was observed in some infantile patients (up to 22 mumol/L) compared with age-matched controls (mean, 2.2 mumol/L). The method. . . determine oligosaccharide concentrations in a single 3-mm blood spot, but no differences were observed between blood spots from control and **Pompe**-affected individuals. Conclusions: Measurements of oligosaccharide concentrations in urine by this new method have potential application for the diagnosis and monitoring of patients with **Pompe** disease. Plasma analysis may have limited application for infantile patients, but analysis of blood spots does not discriminate between controls. . .
IT . . .
IT Parts, Structures, & Systems of Organisms
blood: blood and lymphatics; plasma: blood and lymphatics; urine: excretory system
IT Diseases
Pompe disease: genetic disease, metabolic disease
Glycogen Storage Disease Type II (MeSH)
IT Diseases
lysosomal storage disorder: metabolic disease
IT Chemicals & Biochemicals
1-phenyl-3-methyl-5-pyrazolone; creatinine; oligosaccharides: determination; **tetrasaccharide**

L3 ANSWER 3 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
TI Validation of glucose **tetrasaccharide** as a biomarker for diagnosis and monitoring enzyme replacement therapy for **Pompe** disease.
IT Major Concepts
Genetics
IT Parts, Structures, & Systems of Organisms
blood: blood and lymphatics; urine: excretory system
IT Diseases
Pompe disease: genetic disease, metabolic disease
Glycogen Storage Disease Type II (MeSH)
IT Chemicals & Biochemicals
acid a-glucosidase; glucose **tetrasaccharide**: biomarker

L3 ANSWER 4 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
TI Liquid chromatographic assay for a glucose **tetrasaccharide**, a putative biomarker for the diagnosis of **Pompe** disease.
AB. . . tetraglucose, normally excreted in the urine, has previously been shown to be elevated in a number of pathological conditions including

Pompe disease (glycogen storage disease type II), which is caused by a deficiency of the lysosomal enzyme acid alpha-glucosidase. Concentrations of. . . 1-5, 6-10, 11-20, and >20 years, both in normal individuals and in a cohort of 21 patients with enzymatically confirmed **Pompe** disease. The Glc4 concentration decreased with age in both groups, but all the patients had elevated Glc4 levels compared with. . . urine. Our results demonstrate that this method is suitable for application in clinical laboratories to help establish the diagnosis of **Pompe** disease.

IT Major Concepts
Clinical Chemistry (Allied Medical Sciences); Methods and Techniques

IT Diseases
Pompe disease: genetic disease, metabolic disease
Glycogen Storage Disease Type II (MeSH)

IT Chemicals & Biochemicals
glucose **tetrasaccharide**: assay, biomarker

L3 ANSWER 5 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN

AB. . . characterization and some properties of leucocyte α-glucosidase towards disaccharides with the α-1, 4 (maltose) and α-1, 6-glucosidic linkage (isomaltose) and **tetrasaccharides** with the α-1, 4 (maltotetraose) and α-1,6-i glucosidic linkage (**tetrasaccharide**, Glcαl → 6Glcαl → 4Glcαl → 4Glc, which was isolated from the urine of a patient with glycogenosis type II).. . . neutral pH. Acid α-glucosidase could hydrolyze maltose about 10 times faster than isomaltose, and maltotetraose about 5 times faster than **tetrasaccharide** isolated from urine. In leucocytes of the patient with late onset glycogenosis type II, acid α-glucosidase activities towards maltose, isomaltose, maltotetraose and **tetrasaccharide** isolated from urine showed 75.3%, 67.4%, 76.5% and 41.4% of normal control values, respectively. Neutral α-glucosidase activities towards these four oligosaccharides were normal. **Tetrasaccharide** with α-1, 6-glucosidic linkage might be accumulated by the impaired hydrolysis in the circulation as well as the leakage of. . .

IT Miscellaneous Descriptors
HUMAN MUSCLE **POMPE**'S DISEASE

=> d l3 iall 1-5

L3 ANSWER 1 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN

ACCESSION NUMBER: 2003:298793 BIOSIS
DOCUMENT NUMBER: PREV200300298793
TITLE: Analysis of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry.
AUTHOR(S): Young, Sarah P. [Reprint Author]; Stevens, Robert D.; An, Yan; Chen, Yuan-Tsong; Millington, David S.
CORPORATE SOURCE: Biochemical Genetics Laboratory, Division of Medical Genetics, Department of Pediatrics, Duke University Medical Center, 99 TW Alexander Drive, P.O. Box 14991, Research Triangle Park, NC, 27709, USA
syoung@duke.edu
SOURCE: Analytical Biochemistry, (May 15 2003) Vol. 316, No. 2, pp. 175-180. print.
ISSN: 0003-2697 (ISSN print).
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 25 Jun 2003
Last Updated on STN: 1 Aug 2003

ABSTRACT:Patients with glycogen storage disease type II (GSD II) typically excrete increased amounts of a glycogen-derived glucose **tetrasaccharide** , Glcalpha1-6Glcalpha1-4Glcalpha1-4Glc (Glc4), in the urine. With the advent of a new enzyme replacement therapy for GSD II, there is a need for early

identification of patients with this disease and for monitoring the efficacy of treatment. Glc4 is a good candidate biomarker for GSD II. A simple and robust method using stable isotope dilution-electrospray ionization-tandem mass spectrometry for the analysis of Glc4 in biological samples was developed. A 13C6-labeled stable isotope internal standard was synthesized by transglycosylation using a recombinant alpha-amylase. Butyl 4-aminobenzoate derivatives of Glc4 and the internal standard were analyzed using multiple reaction monitoring. This method was shown to be accurate and precise by the repeated analysis of calibrators and quality control samples in urine and plasma. There was good agreement with a high-performance liquid chromatography-UV method for urine samples, whereas there was less agreement with plasma samples. Accurate determination from dried urine spot samples was also demonstrated. This method is amenable to high-throughput analysis, a necessary prerequisite for mass screening for GSD II.

CONCEPT CODE: Genetics - Human 03508
Biochemistry studies - General 10060
Biochemistry studies - Carbohydrates 10068
Pathology - Therapy 12512
Metabolism - General metabolism and metabolic pathways 13002
Metabolism - Metabolic disorders 13020
Blood - Blood and lymph studies 15002
Blood - Blood cell studies 15004
Urinary system - Physiology and biochemistry 15504

INDEX TERMS: Major Concepts
Biochemistry and Molecular Biophysics; Medical Genetics (Allied Medical Sciences); Metabolism; Methods and Techniques

INDEX TERMS: Parts, Structures, & Systems of Organisms
plasma: blood and lymphatics; urine: excretory system

INDEX TERMS: Diseases
Pompe disease: genetic disease, metabolic disease
Glycogen Storage Disease Type II (MeSH)

INDEX TERMS: Diseases
glycogen storage disease type II: genetic disease, metabolic disease, therapy, GSD II
Glycogen Storage Disease Type II (MeSH)

INDEX TERMS: Chemicals & Biochemicals
butyl 4-aminobenzoate derivatives; glucose; glucose **tetrasaccharide**: analysis; recombinant alpha-amylase

INDEX TERMS: Methods & Equipment
enzyme replacement therapy: clinical techniques, therapeutic and prophylactic techniques; high performance liquid chromatography-UV method: chromatographic techniques, laboratory techniques; high-throughput analysis: laboratory techniques; multiple reaction monitoring: laboratory techniques; stable isotope dilution-electrospray ionization tandem mass spectrometry: laboratory techniques, spectrum analysis techniques; transglycosylation: laboratory techniques

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human (common): patient
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

REGISTRY NUMBER: 94-25-7D (butyl 4-aminobenzoate derivatives)
50-99-7Q (glucose)
58367-01-4Q (glucose)

L3 ANSWER 2 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2002:178595 BIOSIS
DOCUMENT NUMBER: PREV200200178595
TITLE: Determination of oligosaccharides in **Pompe** disease by electrospray ionization tandem mass spectrometry.
AUTHOR(S): Rozaklis, Tina; Ramsay, Steven L.; Whitfield, Phillip D.; Ranieri, Enzo; Hopwood, John J.; Meikle, Peter J. [Reprint author]
CORPORATE SOURCE: Lysosomal Diseases Research Unit, Department of Chemical Pathology, Women's and Children's Hospital, 72 King William Road, North Adelaide, South Australia, 5006, Australia peter.meikle@adelaide.edu.au
SOURCE: Clinical Chemistry, (January, 2002) Vol. 48, No. 1, pp. 131-139. print.
CODEN: CLCHAU. ISSN: 0009-9147.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 6 Mar 2002
Last Updated on STN: 6 Mar 2002

ABSTRACT:Background: The development of therapies for lysosomal storage disorders has created a need for biochemical markers to monitor the efficacy of therapy and methods to quantify these markers in biologic samples. In ***Pompe*** disease, the concentration of a **tetrasaccharide**, consisting of four glucose residues, is reputedly increased in urine and plasma, but faster and more sensitive methods are required for the analysis of this, and other oligosaccharides, from biologic fluids. Methods: We optimized the derivatization of storage oligosaccharides with 1-phenyl-3-methyl-5-pyrazolone for the measurement, by electrospray ionization tandem mass spectrometry, of oligosaccharide concentrations in urine (n = 6), plasma (n = 11), and dried-blood spots (n = 17) from **Pompe**-affected individuals. Age-matched control samples of urine (n = 10), plasma (n = 28), and blood spots (n = 369) were also analyzed. Results: The mean **tetrasaccharide** concentration was increased in urine from infantile-onset (0.69-12 mmol/mol of creatinine) and adult-onset (0.22-3.0 mmol/mol of creatinine) **Pompe** individuals compared with age-matched controls. In plasma samples, an increased **tetrasaccharide** concentration was observed in some infantile patients (up to 22 mumol/L) compared with age-matched controls (mean, 2.2 mumol/L). The method developed was sensitive enough to determine oligosaccharide concentrations in a single 3-mm blood spot, but no differences were observed between blood spots from control and **Pompe**-affected individuals. Conclusions: Measurements of oligosaccharide concentrations in urine by this new method have potential application for the diagnosis and monitoring of patients with **Pompe** disease. Plasma analysis may have limited application for infantile patients, but analysis of blood spots does not discriminate between controls and affected individuals.

CONCEPT CODE: Genetics - Human 03508
Clinical biochemistry - General methods and applications 10006
Biochemistry studies - Proteins, peptides and amino acids 10064
Biochemistry studies - Carbohydrates 10068
Metabolism - Metabolic disorders 13020
Blood - Blood and lymph studies 15002
Blood - Blood cell studies 15004
Urinary system - Physiology and biochemistry 15504
Pediatrics - 25000

INDEX TERMS: Major Concepts
Clinical Chemistry (Allied Medical Sciences); Medical Genetics (Allied Medical Sciences); Methods and Techniques

INDEX TERMS: Parts, Structures, & Systems of Organisms
blood: blood and lymphatics; plasma: blood and lymphatics; urine: excretory system

INDEX TERMS: Diseases
Pompe disease: genetic disease, metabolic disease
Glycogen Storage Disease Type II (MeSH)
INDEX TERMS: Diseases
lysosomal storage disorder: metabolic disease
INDEX TERMS: Chemicals & Biochemicals
1-phenyl-3-methyl-5-pyrazolone; creatinine; oligosaccharides: determination; **tetrasaccharide**
INDEX TERMS: Methods & Equipment
electrospray ionization tandem mass spectrometry: determination method; plasma analysis: analytical method
ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human: adult, child, infant, patient
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates
REGISTRY NUMBER: 89-25-8Q (1-phenyl-3-methyl-5-pyrazolone)
19735-89-8Q (1-phenyl-3-methyl-5-pyrazolone)
60-27-5 (creatinine)

L3 ANSWER 3 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2001:554213 BIOSIS
DOCUMENT NUMBER: PREV200100554213
TITLE: Validation of glucose **tetrasaccharide** as a biomarker for diagnosis and monitoring enzyme replacement therapy for **Pompe** disease.
AUTHOR(S): An, Y. [Reprint author]; Millington, D. S. [Reprint author]; Kishnani, P. [Reprint author]; Amalfitano, A. [Reprint author]; Chen, Y. T. [Reprint author]
CORPORATE SOURCE: Division of Medical Genetics, Department of Pediatrics, Duke Univ Medical Center, Durham, NC, USA
SOURCE: American Journal of Human Genetics, (October, 2001) Vol. 69, No. 4 Supplement, pp. 482. print.
Meeting Info.: 51st Annual Meeting of the American Society of Human Genetics. San Diego, California, USA. October 12-16, 2001.
CODEN: AJHGAG. ISSN: 0002-9297.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
Conference; (Meeting Poster)
LANGUAGE: English
ENTRY DATE: Entered STN: 28 Nov 2001
Last Updated on STN: 25 Feb 2002
CONCEPT CODE: General biology - Symposia, transactions and proceedings 00520
Genetics - General 03502
Genetics - Animal 03506
Pathology - Therapy 12512
Metabolism - Metabolic disorders 13020
Blood - Blood and lymph studies 15002
Blood - Blood cell studies 15004
Urinary system - Physiology and biochemistry 15504
INDEX TERMS: Major Concepts
Genetics
INDEX TERMS: Parts, Structures, & Systems of Organisms
blood: blood and lymphatics; urine: excretory system
INDEX TERMS: Diseases
Pompe disease: genetic disease, metabolic disease
Glycogen Storage Disease Type II (MeSH)

INDEX TERMS: Chemicals & Biochemicals
acid a-glucosidase; glucose **tetrasaccharide**: biomarker
INDEX TERMS: Methods & Equipment
enzyme replacement therapy: therapeutic method
INDEX TERMS: Miscellaneous Descriptors
Meeting Abstract; Meeting Poster
ORGANISM: Classifier
Galliformes 85536
Super Taxa
Aves; Vertebrata; Chordata; Animalia
Organism Name
quail: animal model
Taxa Notes
Animals, Birds, Chordates, Nonhuman Vertebrates, Vertebrates

L3 ANSWER 4 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 2001:26713 BIOSIS
DOCUMENT NUMBER: PREV200100026713
TITLE: Liquid chromatographic assay for a glucose **tetrasaccharide**, a putative biomarker for the diagnosis of **Pompe** disease.
AUTHOR(S): An, Yan; Young, Sarah P.; Hillman, Stephen L.; Van Hove, Johan L. K. [Reprint author]; Chen, Yuan-Tsong; Millington, David S.
CORPORATE SOURCE: Division of Medical Genetics, Department of Pediatrics, Duke University Medical Center, 99 TW Alexander Drive, Research Triangle Park, NC, 27709, USA
vanan@acpub.duke.edu
SOURCE: Analytical Biochemistry, (December 1, 2000) Vol. 287, No. 1, pp. 136-143. print.
CODEN: ANBCA2. ISSN: 0003-2697.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 10 Jan 2001
Last Updated on STN: 12 Feb 2002

ABSTRACT:A HPLC method associated with butyl-p-aminobenzoate derivatization has been developed for the analysis of a tetraglucose oligomer, Glcalpha1-6Glcalpha1-4Glcalpha1-4Glc, designated Glc4, in biological fluids. This tetraglucose, normally excreted in the urine, has previously been shown to be elevated in a number of pathological conditions including **Pompe** disease (glycogen storage disease type II), which is caused by a deficiency of the lysosomal enzyme acid alpha-glucosidase. Concentrations of Glc4 in both urine and plasma were established for the age ranges of <1, 1-5, 6-10, 11-20, and >20 years, both in normal individuals and in a cohort of 21 patients with enzymatically confirmed **Pompe** disease. The Glc4 concentration decreased with age in both groups, but all the patients had elevated Glc4 levels compared with age-matched controls. Electrospray tandem mass spectrometry was employed to establish the homogeneity of the HPLC peak for Glc4 and to investigate the identity of other unusual oligosaccharides excreted in patient urine. Our results demonstrate that this method is suitable for application in clinical laboratories to help establish the diagnosis of ***Pompe*** disease.

CONCEPT CODE: Pathology - Diagnostic 12504
Genetics - Human 03508
Clinical biochemistry - General methods and applications 10006
Metabolism - Metabolic disorders 13020
INDEX TERMS: Major Concepts
Clinical Chemistry (Allied Medical Sciences); Methods and Techniques
INDEX TERMS: Diseases
Pompe disease: genetic disease, metabolic disease

Glycogen Storage Disease Type II (MeSH)

INDEX TERMS: Chemicals & Biochemicals
glucose **tetrasaccharide**: assay, biomarker

INDEX TERMS: Methods & Equipment
electrospray tandem mass spectrometry: analytical method, mass spectrometry: CB; high performance liquid chromatography [HPLC]: diagnostic method, liquid chromatography

ORGANISM: Classifier
Hominidae 86215
Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia
Organism Name
human
Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

L3 ANSWER 5 OF 5 BIOSIS COPYRIGHT 2004 BIOLOGICAL ABSTRACTS INC. on STN
ACCESSION NUMBER: 1991:6034 BIOSIS
DOCUMENT NUMBER: PREV199191006034; BA91:6034
TITLE: LEUKOCYTE ALPHA-1 4 GLUCOSIDASE AND ALPHA-1 6 GLUCOSIDASE ACTIVITIES TOWARDS OLIGOSACCHARIDES IN LATE ONSET GLYCOGENOSIS TYPE II.
AUTHOR(S): KURIYAMA M [Reprint author]; HIWATARI R-I; OSAME M; IGATA A
CORPORATE SOURCE: THIRD DEP INTERN MED, KAGOSHIMA UNIV SCH MED, KAGOSHIMA 890, JPN
SOURCE: Tohoku Journal of Experimental Medicine, (1990) Vol. 161, No. 4, pp. 343-351.
CODEN: TJEMAO. ISSN: 0040-8727.
DOCUMENT TYPE: Article
FILE SEGMENT: BA
LANGUAGE: ENGLISH
ENTRY DATE: Entered STN: 8 Dec 1990
Last Updated on STN: 30 Jan 1991

ABSTRACT:We describe the partial characterization and some properties of leucocyte α-glucosidase towards disaccharides with the α-1, 4 (maltose) and α-1, 6-glucosidic linkage (isomaltose) and ***tetrasaccharides*** with the α-1, 4 (maltotetraose) and α-1,6-i glucosidic linkage (**tetrasaccharide**, Glcα1 → 6Glcα1 → 4Glcα1 → 4Glc, which was isolated from the urine of a patient with glycogenosis type II). Leucocyte α-glucosidase showed optimal activity towards all four oligosaccharides under two conditions, acidic (pH 4.0-4.5) and neutral (pH 6.0-6.5) regions. Our comparative studies on enzyme kinetics showed that leucocyte α-glucosidase was able to hydrolyze both the 1 → 4 isomers and the 1 → 6 isomers at acidic and neutral pH. Acid α-glucosidase could hydrolyze maltose about 10 times faster than isomaltose, and maltotetraose about 5 times faster than **tetrasaccharide** isolated from urine. In leucocytes of the patient with late onset glycogenosis type II, acid α-glucosidase activities towards maltose, isomaltose, maltotetraose and ***tetrasaccharide*** isolated from urine showed 75.3%, 67.4%, 76.5% and 41.4% of normal control values, respectively. Neutral α-glucosidase activities towards these four oligosaccharides were normal. ***Tetrasaccharide*** with α-1, 6-glucosidic linkage might be accumulated by the impaired hydrolysis in the circulation as well as the leakage of undegraded glycogen to the circulation from the affected muscle.

CONCEPT CODE: Genetics - Human 03508
Clinical biochemistry - General methods and applications 10006
Biochemistry studies - Carbohydrates 10068
Metabolism - Carbohydrates 13004
Metabolism - Metabolic disorders 13020
Blood - Blood cell studies 15004
Blood - Lymphatic tissue and reticuloendothelial system

15008
Muscle - Pathology 17506

INDEX TERMS: Major Concepts
Blood and Lymphatics (Transport and Circulation); Clinical Chemistry (Allied Medical Sciences); Metabolism; Muscular System (Movement and Support)

INDEX TERMS: Miscellaneous Descriptors
HUMAN MUSCLE **POMPE**'S DISEASE

ORGANISM: Classifier
Hominidae 86215

Super Taxa
Primates; Mammalia; Vertebrata; Chordata; Animalia

Taxa Notes
Animals, Chordates, Humans, Mammals, Primates, Vertebrates

REGISTRY NUMBER: 9001-42-7 (ALPHA-1,4-GLUCOSIDASE)
37288-48-5 (ALPHA-1,6-GLUCOSIDASE)

```
=> file caplus
COST IN U.S. DOLLARS                          SINCE FILE      TOTAL
                                                   ENTRY    SESSION
FULL ESTIMATED COST                               27.10      27.31

FILE 'CAPLUS' ENTERED AT 19:21:46 ON 10 MAR 2004
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```
FILE COVERS 1907 - 10 Mar 2004  VOL 140 ISS 11
FILE LAST UPDATED: 9 Mar 2004  (20040309/ED)

  This file contains CAS Registry Numbers for easy and accurate
  substance identification.


=> s pompe
           248 POMPE
            15 POMPES
L4         250 POMPE
                 (POMPE OR POMPES)

=> s l4 and (tetrasaccharide)
          2456 TETRASACCHARIDE
           850 TETRASACCHARIDES
          2996 TETRASACCHARIDE
                 (TETRASACCHARIDE OR TETRASACCHARIDES)
L5           8 L4 AND (TETRASACCHARIDE)

=> d l5 kwic 1-8

L5   ANSWER 1 OF 8  CAPLUS  COPYRIGHT 2004 ACS on STN
TI   Analysis of a glucose tetrasaccharide elevated in Pompe
     disease by stable isotope dilution-electrospray ionization tandem mass
```

spectrometry
AB Patients with glycogen storage disease type II (GSD II) typically excrete increased amts. of a glycogen-derived glucose **tetrasaccharide**, Glcα1-6Glcα1-4Glcα1-4Glc (Glc4), in the urine. With the advent of a new enzyme replacement therapy for GSD II, there is a. . .
ST glucose **tetrasaccharide** mass spectrometry glycogen storage disease urine
IT Blood analysis
Blood plasma
HPLC
Human
Isotope dilution mass spectrometry
Quality control
Urine analysis
(anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)
IT Tandem mass spectrometry
(electrospray-ionization; anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)
IT Electrospray ionization mass spectrometry
(tandem; anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)
IT Glycogen storage disease
(type II; anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)
IT 35175-16-7
RL: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)

L5 ANSWER 2 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
TI Determination of oligosaccharides in **pompe** disease by electrospray ionization tandem mass spectrometry
AB . . . need for biochem. markers to monitor the efficacy of therapy and methods to quantify these markers in biol. samples. In **Pompe** disease, the concentration of a **tetrasaccharide**, consisting of four glucose residues, is reputedly increased in urine and plasma, but faster and more sensitive methods are required. . . spectrometry, of oligosaccharide concns. in urine (n = 6), plasma (n = 11), and dried-blood spots (n = 17) from **Pompe**-affected individuals. Age-matched control samples of urine (n = 10), plasma (n = 28), and blood spots (n = 369) were also analyzed. Results: The mean **tetrasaccharide** concentration was increased in urine from infantile-onset (0.69-12 mmol/ mol. of
creatinine) and adult-onset (0.22-3.0 mmolfmol of creatinine) **Pompe** individuals compared with age-matched controls. In plasma samples, an increased **tetrasaccharide** concentration was observed in some infantile patients (up to 22 μmol/L) compared with age-matched controls (mean, 2.2 μmol/L). The method. . . determine oligosaccharide concns. in a single 3-mm blood spot, but no differences were observed between blood spots from control and **Pompe**-affected individuals. Conclusions: Measurements of oligosaccharide concns. in urine by this new method have potential application for the diagnosis and monitoring of patients with **Pompe** disease. Plasma anal. may have limited application for infantile patients, but anal. of blood spots does not discriminate between controls. . .
ST oligosaccharide **pompe** disease electrospray ionization tandem mass spectrometry
IT Blood analysis

Blood plasma
Body fluid
Diagnosis
Human
Tandem mass spectrometry
Urine analysis
(oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

IT Oligosaccharides, analysis
RL: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

IT Glycogen storage disease
(type II; oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

IT 89-25-8, 1-Phenyl-3-methyl-5-pyrazolone
RL: BUU (Biological use, unclassified); BIOL (Biological study); USES (Uses)
(oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

L5 ANSWER 3 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN

TI Diagnostic methods for **Pompe** disease and other glycogen storage diseases

AB The invention concerns methods of screening subjects for lysosomal storage diseases, preferably glycogen storage diseases, using a **tetrasaccharide** as a biomarker. In a more preferred embodiment, subjects are screened for **Pompe** disease (i.e., glycogen storage disease type II). Also provided are neonatal screening assays. The present invention further provides methods of monitoring the clin. condition and efficacy of therapeutic treatment in affected subjects. Further provided are methods of measuring a **tetrasaccharide** biomarker by tandem mass spectrometry, preferably, as part of a neonatal screening assay for **Pompe** disease.

ST glycogen storage **Pompe** disease diagnosis **tetrasaccharide** HPLC mass spectrometry

IT Amniotic fluid
Animal tissue
Biomarkers (biological responses)
Blood analysis
Blood plasma
Blood serum
Body fluid
Cell
Electrospray ionization mass spectrometry
Glycogen storage disease
HPLC
Human
Immunoassay
Liquid chromatography
Lysosomal storage disease
Mass spectrometry
Newborn
Purification
Sputum
Tandem mass spectrometry
Urine analysis
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)

IT Blood analysis
(dried blood spot; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

IT Standard substances, analytical
(internal; diagnostic methods for **Pompe** disease and other

glycogen storage diseases)
IT Ecology
(population; diagnostic methods for **Pompe** disease and other glycogen storage diseases)
IT Glycogen storage disease
(type II; diagnostic methods for **Pompe** disease and other glycogen storage diseases)
IT Glycogen storage disease
(type III; diagnostic methods for **Pompe** disease and other glycogen storage diseases)
IT Glycogen storage disease
(type VI; diagnostic methods for **Pompe** disease and other glycogen storage diseases)
IT 35175-16-7 379261-84-4
RL: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)
IT 89-25-8, 1-Phenyl-3-methyl-5-pyrazolone 93-97-0, Benzoic anhydride 94-25-7, Butyl p-aminobenzoate 27918-14-5, 2-Aminoacridone
RL: ARG (Analytical reagent use); ANST (Analytical study); USES (Uses)
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)
IT 34620-76-3, Maltopentaose 34620-77-4, Maltohexaose 379261-83-3
RL: ARU (Analytical role, unclassified); ANST (Analytical study)
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)

L5 ANSWER 4 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
TI Liquid chromatographic assay for a glucose **tetrasaccharide**, a putative biomarker for the diagnosis of **pompe** disease
AB . . . tetraglucose, normally excreted in the urine, has previously been shown to be elevated in a number of pathol. conditions including **Pompe** disease (glycogen storage disease type II), which is caused by a deficiency of the lysosomal enzyme acid α-glucosidase. Concns. of. . . . 1-5, 6-10, 11-20, and >20 yr, both in normal individuals and in a cohort of 21 patients with enzymically confirmed **Pompe** disease. The Glc4 concentration decreased with age in both groups, but all the patients had elevated Glc4 levels compared with. . . urine. Our results demonstrate that this method is suitable for application in clin. labs. to help establish the diagnosis of **Pompe** disease. (c) 2000 Academic Press.
ST glucose **tetrasaccharide** detn HPLC **Pompe** disease
IT HPLC
Urine analysis
(glucose **tetrasaccharide** detn by HPLC for diagnosis of **Pompe** disease)
IT Glycogen storage disease
(type II; glucose **tetrasaccharide** detn by HPLC for diagnosis of **Pompe** disease)
IT 35175-16-7
RL: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(tetraglucose; glucose **tetrasaccharide** detn by HPLC for diagnosis of **Pompe** disease)

L5 ANSWER 5 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
AB . . . the partial characterization and some properties of leukocyte α-glucosidase towards disaccharides with the α-1,4 (maltose) and α-1,6-glucosidic linkage (isomaltose) and **tetrasaccharides** with the α-1,4 (maltotetraose) and α-1,6-glucosidic linkage (**tetrasaccharide**, Glcα1→6Glcα1→4Glcα1→4Glc, which was isolated from the urine of a patient with glycogenosis type II). Leukocyte α-glucosidase showed optimal activity towards. . .

neutral pH. Acid α-glucosidase could hydrolyze maltose about 10 times faster than isomaltose, and maltotetraose about 5 times faster than **tetrasaccharide** isolated from urine. In leukocytes of the patient with late onset glycogenosis type II, acid α-glucosidase activities towards maltose, isomaltose, maltotetraose and **tetrasaccharide** isolated from urine showed 75.3%, 67.4%, 76.5% and 41.4% of normal control values, resp. Neutral α-glucosidase activities towards these four oligosaccharides were normal. **Tetrasaccharide** with α-1,6-glucosidic linkage might be accumulated by the impaired hydrolysis in the circulation as well as the leakage of undergraded. . .

IT Leukocyte
(α-glucosidase of, in **Pompe**'s disease in human, oligosaccharides hydrolysis by)

IT Glycogenosis
(**Pompe**'s disease, α-glucosidase of leukocytes in, in human, oligosaccharides hydrolysis by)

L5 ANSWER 6 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN

AB . . . ..., are specifically bound by the antibody with similar affinities, but the affinity is somewhat higher for chains containing the **tetrasaccharide** sequence Glcα1-6Glcα1-4Glcα1-4Glc at the nonreducing terminal. Utilization of affinity methods offers clear advantages for isolation and characterization of oligosaccharides with. . .

IT Glycogenosis
(**Pompe**'s disease, oligodextrins of urine in, structural and immunochem. anal. of)

L5 ANSWER 7 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN

TI Thin-layer chromatography of oligosaccharides in urine as a rapid indication for the diagnosis of lysosomal acid maltase deficiency (**Pompe**'s disease)

AB . . . desalted by ion-exchange chromatog. and then analyzed by the TLC method of R. Humbel and M. Collart (1975). The glucose-containing **tetrasaccharide** α-D-Glc-(1→6)-α-D-Glc-(1→4)-α-D-Glc-(1→4)-D-Glc was detected in urine of patients with **Pompe**'s disease. This method is potentially useful for the rapid and reliable diagnosis of lysosomal acid maltase deficiency. However, lactobionic acid. . .

ST urine oligosaccharide thin layer chromatog; **Pompe** disease diagnosis oligosaccharide TLC; lysosomal acid maltase deficiency diagnosis; desalting urine oligosaccharide TLC

IT Urine analysis
(oligosaccharides detection in, of humans by TLC in **Pompe**'s disease diagnosis, desalting in)

IT Glycogenosis
(**Pompe**'s disease, diagnosis of, oligosaccharides of human urine in)

IT 35175-16-7
RL: ANT (Analyte); ANST (Analytical study)
(detection of, in human urine by TLC in **Pompe**'s disease diagnosis)

IT 96-82-2
RL: ANST (Analytical study)
(interference by, in oligosaccharides detection in human urine by TLC in **Pompe**'s disease diagnosis)

L5 ANSWER 8 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN

TI Increased excretion of a glucose-containing **tetrasaccharide** in the urine of a patient with glycogen storage disease type II (**Pompe**'s disease)

AB A **tetrasaccharide** was isolated from the urine (13 mg/24 hr) of a 10-year-old boy with the childhood form of the title disease.. . . amts. from normal urine. Larger glucose-containing oligosaccharides, not detected in normal urine, were also present in the urine of the

Pompe case.
ST **Pompe** disease carbohydrate structure; glycogen storage disease **tetrasaccharide** urine; urine glycogen storage disease
IT Glycogenosis
(glucose-containing **tetrasaccharide** of urine in)

=> d l5 iall 1-8

L5 ANSWER 1 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 2003:311407 CAPLUS
DOCUMENT NUMBER: 140:90078
ENTRY DATE: Entered STN: 23 Apr 2003
TITLE: Analysis of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry
AUTHOR(S): Young, Sarah P.; Stevens, Robert D.; An, Yan; Chen, Yuan-Tsong; Millington, David S.
CORPORATE SOURCE: Department of Pediatrics, Division of Medical Genetics, Biochemical Genetics Laboratory, Duke University Medical Center, Research Triangle Park, NC, 27709, USA
SOURCE: Analytical Biochemistry (2003), 316(2), 175-180
CODEN: ANBCA2; ISSN: 0003-2697
PUBLISHER: Elsevier Science
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 9-5 (Biochemical Methods)
Section cross-reference(s): 1

ABSTRACT:
Patients with glycogen storage disease type II (GSD II) typically excrete increased amts. of a glycogen-derived glucose **tetrasaccharide**, Glcα1-6Glcα1-4Glcα1-4Glc (Glc4), in the urine. With the advent of a new enzyme replacement therapy for GSD II, there is a need for early identification of patients with this disease and for monitoring the efficacy of treatment. Glc4 is a good candidate biomarker for GSD II. A simple and robust method using stable isotope dilution-electrospray ionization-tandem mass spectrometry for the anal. of Glc4 in biol. samples was developed. A 13C6-labeled stable isotope internal standard was synthesized by transglycosylation using a recombinant α-amylase. Bu 4-aminobenzoate derivs. of Glc4 and the internal standard were analyzed using multiple reaction monitoring. This method was shown to be accurate and precise by the repeated anal. of calibrators and quality control samples in urine and plasma. There was good agreement with a high-performance liquid chromatog.-UV method for urine samples, whereas there was less agreement with plasma samples. Accurate determination from dried urine spot samples was also demonstrated. This method is amenable to high-throughput anal., a necessary prerequisite for mass screening for GSD II.

SUPPL. TERM: glucose **tetrasaccharide** mass spectrometry glycogen storage disease urine
INDEX TERM: Blood analysis
Blood plasma
HPLC
Human
Isotope dilution mass spectrometry
Quality control
Urine analysis
(anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)
INDEX TERM: Tandem mass spectrometry
(electrospray-ionization; anal. of a glucose

tetrasaccharide elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)

INDEX TERM: Electrospray ionization mass spectrometry (tandem; anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)

INDEX TERM: Glycogen storage disease (type II; anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)

INDEX TERM: 35175-16-7
ROLE: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(anal. of a glucose **tetrasaccharide** elevated in **Pompe** disease by stable isotope dilution-electrospray ionization tandem mass spectrometry)

REFERENCE COUNT: 18 THERE ARE 18 CITED REFERENCES AVAILABLE FOR THIS RECORD.

REFERENCE(S):
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(5) Hallgren, P; Eur J Clin Invest 1974, V4, P429 CAPLUS
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L5 ANSWER 2 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN

ACCESSION NUMBER: 2002:15925 CAPLUS
DOCUMENT NUMBER: 136:131060
ENTRY DATE: Entered STN: 08 Jan 2002

TITLE: Determination of oligosaccharides in **pompe** disease by electrospray ionization tandem mass spectrometry

AUTHOR(S): Rozaklis, Tina; Ramsay, Steven L.; Whitfield, Phillip D.; Ranieri, Enzo; Hopwood, John J.; Meikle, Peter J.

CORPORATE SOURCE: Lysosomal Diseases Research Unit, Women's and Children's Hospital, North Adelaide, South Australia, 5006, Australia

SOURCE: Clinical Chemistry (Washington, DC, United States) (2002), 48(1), 131-139
CODEN: CLCHAU; ISSN: 0009-9147

PUBLISHER: American Association for Clinical Chemistry
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 9-5 (Biochemical Methods)
Section cross-reference(s): 14

ABSTRACT:
Background: The development of therapies for lysosomal storage disorders has created a need for biochem. markers to monitor the efficacy of therapy and methods to quantify these markers in biol. samples. In **Pompe** disease, the concentration of a **tetrasaccharide**, consisting of four glucose residues, is reputedly increased in urine and plasma, but faster and more sensitive methods are required for the anal. of this, and other

oligosaccharides, from biol. fluids. Methods: We optimized the derivatization of storage oligosaccharides with 1-phenyl-3-methyl-5-pyrazolone for the measurement, by electrospray ionization tandem mass spectrometry, of oligosaccharide concns. in urine (n = 6), plasma (n = 11), and dried-blood spots (n = 17) from **Pompe**-affected individuals. Age-matched control samples of urine (n = 10), plasma (n = 28), and blood spots (n = 369) were also analyzed. Results: The mean **tetrasaccharide** concentration was increased in urine from infantile-onset (0.69-12 mmol/ mol. of creatinine) and adult-onset (0.22-3.0 mmolfmol of creatinine) **Pompe** individuals compared with age-matched controls. In plasma samples, an increased **tetrasaccharide** concentration was observed in some infantile patients (up to 22 μmol/L) compared with
age-matched controls (mean, 2.2 μmol/L). The method developed was sensitive enough to determine oligosaccharide concns. in a single 3-mm blood spot, but no differences were observed between blood spots from control and **Pompe** -affected individuals. Conclusions: Measurements of oligosaccharide concns. in urine by this new method have potential application for the diagnosis and monitoring of patients with **Pompe** disease. Plasma anal. may have limited application for infantile patients, but anal. of blood spots does not discriminate between controls and affected individuals.

SUPPL. TERM: oligosaccharide **pompe** disease electrospray ionization tandem mass spectrometry

INDEX TERM: Blood analysis
Blood plasma
Body fluid
Diagnosis
Human
Tandem mass spectrometry
Urine analysis
(oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

INDEX TERM: Oligosaccharides, analysis
ROLE: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

INDEX TERM: Glycogen storage disease
(type II; oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

INDEX TERM: 89-25-8, 1-Phenyl-3-methyl-5-pyrazolone
ROLE: BUU (Biological use, unclassified); BIOL (Biological study); USES (Uses)
(oligosaccharides determination in **pompe** disease by electrospray ionization tandem mass spectrometry)

REFERENCE COUNT: 28 THERE ARE 28 CITED REFERENCES AVAILABLE FOR THIS RECORD.

REFERENCE(S):

(1) An, Y; Anal Biochem 2000, V287, P136 CAPLUS
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(28) Zaretsky, J; Hum Gene Ther 1997, V8, P1555 CAPLUS

L5 ANSWER 3 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 2001:904726 CAPLUS
DOCUMENT NUMBER: 136:17717
ENTRY DATE: Entered STN: 14 Dec 2001
TITLE: Diagnostic methods for **Pompe** disease and other glycogen storage diseases
INVENTOR(S): Millington, David S.; An, Yan; Chen, Yuan Tsong; Stevens, Robert D.; Young, Sarah P.; Van Hove, Johan L. K.
PATENT ASSIGNEE(S): Duke University, USA; Van Hove, Johan L. K.
SOURCE: PCT Int. Appl., 75 pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent
LANGUAGE: English
INT. PATENT CLASSIF.:
MAIN: G01N033-50
CLASSIFICATION: 9-16 (Biochemical Methods)
Section cross-reference(s): 14
FAMILY ACC. NUM. COUNT: 1
PATENT INFORMATION:

Applicant

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 2001094941	A2	20011213	WO 2001-US18288	20010606
WO 2001094941	A3	20030821		
W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM			
RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG				
US 2002102737	A1	20020801	US 2001-875327	20010606
EP 1360485	A2	20031112	EP 2001-944308	20010606
R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, SE, MC, PT, IE, SI, LT, LV, FI, RO, MK, CY, AL, TR				
JP 2004501365	T2	20040115	JP 2002-502439	20010606

PRIORITY APPLN. INFO.: US 2000-209920P P 20000607
WO 2001-US18288 W 20010606

ABSTRACT:
The invention concerns methods of screening subjects for lysosomal storage diseases, preferably glycogen storage diseases, using a **tetrasaccharide** as a biomarker. In a more preferred embodiment, subjects are screened for ***Pompe*** disease (i.e., glycogen storage disease type II). Also provided are neonatal screening assays. The present invention further provides methods of monitoring the clin. condition and efficacy of therapeutic treatment in affected subjects. Further provided are methods of measuring a ***tetrasaccharide*** biomarker by tandem mass spectrometry, preferably, as part of a neonatal screening assay for **Pompe** disease.

SUPPL. TERM: glycogen storage **Pompe** disease diagnosis **tetrasaccharide** HPLC mass spectrometry

INDEX TERM: Amniotic fluid
Animal tissue
Biomarkers (biological responses)
Blood analysis
Blood plasma
Blood serum
Body fluid
Cell
Electrospray ionization mass spectrometry
Glycogen storage disease
HPLC
Human
Immunoassay
Liquid chromatography
Lysosomal storage disease
Mass spectrometry
Newborn
Purification
Sputum
Tandem mass spectrometry
Urine analysis
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: Blood analysis
(dried blood spot; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: Standard substances, analytical
(internal; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: Ecology
(population; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: Glycogen storage disease
(type II; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: Glycogen storage disease
(type III; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: Glycogen storage disease
(type VI; diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: 35175-16-7 379261-84-4
ROLE: ANT (Analyte); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: 89-25-8, 1-Phenyl-3-methyl-5-pyrazolone 93-97-0, Benzoic anhydride 94-25-7, Butyl p-aminobenzoate 27918-14-5, 2-Aminoacridone
ROLE: ARG (Analytical reagent use); ANST (Analytical study); USES (Uses)
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)

INDEX TERM: 34620-76-3, Maltopentaose 34620-77-4, Maltohexaose 379261-83-3
ROLE: ARU (Analytical role, unclassified); ANST (Analytical study)
(diagnostic methods for **Pompe** disease and other glycogen storage diseases)

L5 ANSWER 4 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 2000:804299 CAPLUS

DOCUMENT NUMBER: 134:175062
ENTRY DATE: Entered STN: 15 Nov 2000
TITLE: Liquid chromatographic assay for a glucose **tetrasaccharide**, a putative biomarker for the diagnosis of **pompe** disease
AUTHOR(S): An, Yan; Young, Sarah P.; Hillman, Stephen L.; Van Hove, Johan L. K.; Chen, Yuan-Tsong; Millington, David S.
CORPORATE SOURCE: Division of Medical Genetics, Department of Pediatrics, Duke University Medical Center, Research Triangle Park, NC, 27709, USA
SOURCE: Analytical Biochemistry (2000), 287(1), 136-143
CODEN: ANBCA2; ISSN: 0003-2697
PUBLISHER: Academic Press
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 9-3 (Biochemical Methods)
Section cross-reference(s): 14

7/6/2000

ABSTRACT:
A HPLC method associated with butyl-p-aminobenzoate derivatization has been developed for the anal. of a tetraglucose oligomer, Glcα1-6Glcα1-4Glcα1-4Glc, designated Glc4, in biol. fluids. This tetraglucose, normally excreted in the urine, has previously been shown to be elevated in a number of pathol. conditions including **Pompe** disease (glycogen storage disease type II), which is caused by a deficiency of the lysosomal enzyme acid α-glucosidase. Concns. of Glc4 in both urine and plasma were established for the age ranges of <1, 1-5, 6-10, 11-20, and >20 yr, both in normal individuals and in a cohort of 21 patients with enzymically confirmed ***Pompe*** disease. The Glc4 concentration decreased with age in both groups, but
all the patients had elevated Glc4 levels compared with age-matched controls. Electrospray tandem mass spectrometry was employed to establish the homogeneity of the HPLC peak for Glc4 and to investigate the identity of other unusual oligosaccharides excreted in patient urine. Our results demonstrate that this method is suitable for application in clin. labs. to help establish the diagnosis of **Pompe** disease. (c) 2000 Academic Press.

SUPPL. TERM: glucose **tetrasaccharide** detn HPLC **Pompe** disease
INDEX TERM: HPLC
Urine analysis
(glucose **tetrasaccharide** detn by HPLC for diagnosis of **Pompe** disease)
INDEX TERM: Glycogen storage disease
(type II; glucose **tetrasaccharide** detn by HPLC for diagnosis of **Pompe** disease)
INDEX TERM: 35175-16-7
ROLE: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
(tetraglucose; glucose **tetrasaccharide** detn by HPLC for diagnosis of **Pompe** disease)
REFERENCE COUNT: 26 THERE ARE 26 CITED REFERENCES AVAILABLE FOR THIS RECORD.
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L5 ANSWER 5 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 1991:59967 CAPLUS
DOCUMENT NUMBER: 114:59967
ENTRY DATE: Entered STN: 23 Feb 1991
TITLE: Leukocyte α-1,4- and α-1,6-glucosidase activities towards oligosaccharides in late onset glycogenosis type II
AUTHOR(S): Kuriyama, Masaru; Hiwatari, Ryoichi; Osame, Mitsuhiro; Igata, Akihiro
CORPORATE SOURCE: Sch. Med., Kagoshima Univ., Kagoshima, 890, Japan
SOURCE: Tohoku Journal of Experimental Medicine (1990), 161(4), 343-51
CODEN: TJEMAO; ISSN: 0040-8727
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 14-14 (Mammalian Pathological Biochemistry)
ABSTRACT:
This study describes the partial characterization and some properties of leukocyte α-glucosidase towards disaccharides with the α-1,4 (maltose) and α-1,6-glucosidic linkage (isomaltose) and ***tetrasaccharides*** with the α-1,4 (maltotetraose) and α-1,6-glucosidic linkage (**tetrasaccharide**, Glcα1→6Glcα1→4Glcα1→4Glc, which was isolated from the urine of a patient with glycogenosis type II). Leukocyte α-glucosidase showed optimal activity towards all four oligosaccharides in acidic (pH 4.0-4.5) and neutral (pH 6.0-6.5) regions. Leukocyte α-glucosidase was able to hydrolyze both the 1→4 isomers and the 1→6 isomers at acidic and neutral pH. Acid α-glucosidase could hydrolyze maltose about 10 times faster than isomaltose, and maltotetraose about 5 times faster than **tetrasaccharide** isolated from urine. In leukocytes of the patient with late onset glycogenosis type II, acid α-glucosidase activities towards maltose, isomaltose, maltotetraose and ***tetrasaccharide*** isolated from urine showed 75.3%, 67.4%, 76.5% and 41.4% of normal control values, resp. Neutral α-glucosidase activities towards these four oligosaccharides were normal. **Tetrasaccharide** with α-1,6-glucosidic linkage might be accumulated by the impaired hydrolysis in the circulation as well as the leakage of undergraded glycogen to the circulation from the affected muscle.

SUPPL. TERM: glycosidase oligosaccharide hydrolysis glycogenosis type II
INDEX TERM: Oligosaccharides
ROLE: RCT (Reactant); RACT (Reactant or reagent)
(hydrolysis of, by α-glucosidase of leukocytes in glycogenosis type II in human)
INDEX TERM: Leukocyte

(α-glucosidase of, in **Pompe**'s disease in human, oligosaccharides hydrolysis by)

INDEX TERM: Glycogenosis
(**Pompe**'s disease, α-glucosidase of leukocytes in, in human, oligosaccharides hydrolysis by)

INDEX TERM: 9001-42-7
ROLE: BIOL (Biological study)
(acid and neutral, of leukocyte in glycogenosis type II in human, oligosaccharides hydrolysis by)

INDEX TERM: 69-79-4, Maltose 499-40-1, Isomaltose 34612-38-9, Maltotetraose 35175-16-7
ROLE: RCT (Reactant); RACT (Reactant or reagent)
(hydrolysis of, by α-glucosidase of leukocytes in glycogenosis type II in human)

L5 ANSWER 6 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 1989:171227 CAPLUS
DOCUMENT NUMBER: 110:171227
ENTRY DATE: Entered STN: 12 May 1989
TITLE: Structural and immunochemical analysis of three α-limit dextrin oligosaccharides
AUTHOR(S): Kumlien, Johan; Groenberg, Gunnar; Nilsson, Bo; Maansson, Olle; Zopf, David; Lundblad, Arne
CORPORATE SOURCE: Dep. Clin. Chem., Univ. Hosp., Lund, S-221 85, Swed.
SOURCE: Archives of Biochemistry and Biophysics (1989), 269(2), 678-89
CODEN: ABBIA4; ISSN: 0003-9861
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 14-14 (Mammalian Pathological Biochemistry)
Section cross-reference(s): 33

ABSTRACT:
Complete structures ae described for 3 urinary oligodextrins from one patient with type II and one patient with type III glycogen storage disease. GLC-MS, direct probe MS, and 1H NMR demonstrate two heptasaccharides and one hexasaccharide containing only α1-4 and α1-6 linkages. The observation that all 3 oligosaccharides were present in urine of both patients and the occurrence of α1-4 and α1-6 linkages in characteristic sequences indicates that the oligodextrins are limit dextrins derived from α-amylolytic degradation of glycogen. The binding affinities of the oligodextrins for a monoclonal antibody (401/6) raised against Glcα1-6Glcα1-4Glcα1-4Glc, were determined by frontal anal. The highest affinity was exhibited by Glcα1-6Glcα1-4Glcα1-4Glc followed by the 2 heptasaccharides with the hexasaccharide. The results from quant. affinity measurements agree with results of structural anal. by phys. methods in that all oligodextrins containing the nonreducing terminal sequence, Glcα1-6Glcα1-4Glc ..., are specifically bound by the antibody with similar affinities, but the affinity is somewhat higher for chains containing the ***tetrasaccharide*** sequence Glcα1-6Glcα1-4Glcα1-4Glc at the nonreducing terminal. Utilization of affinity methods offers clear advantages for isolation and characterization of oligosaccharides with very similar structures.

SUPPL. TERM: dextrin oligosaccharide urine glycogenosis II III

INDEX TERM: Urine
(dextrin oligosaccharide of, in glycogenosis type II and type III in human)

INDEX TERM: Oligosaccharides
ROLE: BIOL (Biological study)
(of α-limit dextrin, of urine in glycogenosis type II and III in human)

INDEX TERM: Glycogenosis
(Cori's disease, oligodextrins of urine in, structural and immunochem. anal. of)

INDEX TERM: Glycogenosis

(**Pompe**'s disease, oligodextrins of urine in, structural and immunochem. anal. of)

INDEX TERM: 9004-53-9, Dextrin 40878-60-2 40878-61-3 40879-32-1
ROLE: PROC (Process)
(of urine, in glycogenosis type II and III in humans, structural and immunochem. anal. of)

L5 ANSWER 7 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 1983:609089 CAPLUS
DOCUMENT NUMBER: 99:209089
ENTRY DATE: Entered STN: 12 May 1984
TITLE: Thin-layer chromatography of oligosaccharides in urine as a rapid indication for the diagnosis of lysosomal acid maltase deficiency (**Pompe**'s disease)
AUTHOR(S): Blom, W.; Luteyn, J. C.; Kelholt-Dijkman, H. H.; Huijmans, J. G. M.; Loonen, M. C. B.
CORPORATE SOURCE: Dep. Pediatr., Univ. Hosp. Rotterdam, Rotterdam, Neth.
SOURCE: Clinica Chimica Acta (1983), 134(1-2), 221-7
CODEN: CCATAR; ISSN: 0009-8981
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 9-3 (Biochemical Methods)
Section cross-reference(s): 14

ABSTRACT:
Urine samples were desalted by ion-exchange chromatog. and then analyzed by the TLC method of R. Humbel and M. Collart (1975). The glucose-containing ***tetrasaccharide*** α-D-Glc-(1→6)-α-D-Glc-(1→4)-α-D-Glc-(1→4)-D-Glc was detected in urine of patients with ***Pompe*** 's disease. This method is potentially useful for the rapid and reliable diagnosis of lysosomal acid maltase deficiency. However, lactobionic acid can interfere with the method.

SUPPL. TERM: urine oligosaccharide thin layer chromatog; **Pompe** disease diagnosis oligosaccharide TLC; lysosomal acid maltase deficiency diagnosis; desalting urine oligosaccharide TLC

INDEX TERM: Chromatography, thin-layer
(of oligosaccharides)

INDEX TERM: Urine analysis
(oligosaccharides detection in, of humans by TLC in **Pompe**'s disease diagnosis, desalting in)

INDEX TERM: Glycogenosis
(**Pompe**'s disease, diagnosis of, oligosaccharides of human urine in)

INDEX TERM: 9001-42-7
ROLE: ANST (Analytical study)
(deficiency of, diagnosis of, oligosaccharides of human urine in)

INDEX TERM: 35175-16-7
ROLE: ANT (Analyte); ANST (Analytical study)
(detection of, in human urine by TLC in **Pompe**'s disease diagnosis)

INDEX TERM: 96-82-2
ROLE: ANST (Analytical study)
(interference by, in oligosaccharides detection in human urine by TLC in **Pompe**'s disease diagnosis)

L5 ANSWER 8 OF 8 CAPLUS COPYRIGHT 2004 ACS on STN
ACCESSION NUMBER: 1975:55516 CAPLUS
DOCUMENT NUMBER: 82:55516
ENTRY DATE: Entered STN: 12 May 1984
TITLE: Increased excretion of a glucose-containing **tetrasaccharide** in the urine of a patient with glycogen storage disease type II (**Pompe**'s disease)

AUTHOR(S): Hallgren, P.; Hansson, G.; Henriksson, K. C.; Hager, A.; Lundblad, Arne; Svensson, S.
CORPORATE SOURCE: Inst. Med. Chem., Univ. Uppsala, Uppsala, Swed.
SOURCE: European Journal of Clinical Investigation (1974), 4(6), 429-33
CODEN: EJCIB8; ISSN: 0014-2972
DOCUMENT TYPE: Journal
LANGUAGE: English
CLASSIFICATION: 14-3 (Mammalian Pathological Biochemistry)
Section cross-reference(s): 13

ABSTRACT:
A **tetrasaccharide** was isolated from the urine (13 mg/24 hr) of a 10-year-old boy with the childhood form of the title disease. The structure, established by sugar anal., methylation anal., optical rotation, and enzymic degradation, was α-D-glucopyranosyl-(1→6)-α-D-glucopyranosyl-(1→4)-α-D-glucopyranosyl-(1→4)-D-glucose. The same compound was also isolated in small amts. from normal urine. Larger glucose-containing oligosaccharides, not detected in normal urine, were also present in the urine of the **Pompe** case.

SUPPL. TERM: **Pompe** disease carbohydrate structure; glycogen storage disease **tetrasaccharide** urine; urine glycogen storage disease
INDEX TERM: Glycogenosis
(glucose-containing **tetrasaccharide** of urine in)
INDEX TERM: Oligosaccharides
ROLE: BIOL (Biological study)
(of urine in glycogen storage disease)
INDEX TERM: 35175-16-7
ROLE: BIOL (Biological study)
(of urine in glucogen storage disease)
INDEX TERM: 25191-16-6
ROLE: BIOL (Biological study)
(oligomeric, of urine in glucogen storage disease)

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COST IN U.S. DOLLARS	SINCE FILE ENTRY	TOTAL SESSION
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